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Questions and Answers about Marfan Syndrome

This booklet answers general questions about Marfan syndrome. It describes the characteristics of the disorder, the diagnostic process, and ways to manage symptoms. If you have additional questions after reading this booklet, you may wish to discuss them with your doctor.

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What Is Marfan Syndrome?

Marfan syndrome is a heritable condition that affects the connective tissue. The primary purpose of connective tissue is to hold the body together and provide a framework for growth and development. In Marfan syndrome, the connective tissue is defective and does not act as it should. Because connective tissue is found throughout the body, Marfan syndrome can affect many body systems, including the skeleton, eyes, heart and blood vessels, nervous system, skin, and lungs. Marfan syndrome affects men, women, and children, and has been found among people of all races and ethnic backgrounds.

What Are the Symptoms of Marfan Syndrome?

Marfan syndrome affects different people in different ways. Some people have only mild symptoms, while others are more severely affected. In most cases, the symptoms progress as the person ages. The body systems most often affected by Marfan syndrome are:

- **Skeleton.** People with Marfan syndrome are typically very tall, slender, and loose-jointed. Because Marfan syndrome affects the long bones of the skeleton, a person's arms, legs, fingers, and toes may be disproportionately long in relation to the rest of the body. A person with Marfan syndrome often has a long, narrow face, and the roof of the mouth may be arched, causing the teeth to be crowded. Other skeletal problems include a sternum (breastbone) that is either protruding or indented, curvature of the spine (scoliosis), and flat feet.
- **Eyes.** More than half of all people with Marfan syndrome experience dislocation of one or both lenses of the eye. The lens may be slightly higher or lower than normal, and may be shifted off to one side. The dislocation may be minimal, or it may be pronounced and obvious. One serious complication that may occur with this disorder is retinal detachment. Many people with Marfan syndrome are also nearsighted (myopic), and some can develop early glaucoma (high pressure within the eye) or cataracts (the eye's lens loses its clearness).
- **Heart and blood vessels (cardiovascular system).** Most people with Marfan syndrome have problems associated with the heart and blood vessels. Because of faulty connective tissue, the wall of the aorta (the large artery that carries blood from the heart to the rest of the body) may be weakened and stretch, a process called aortic dilatation. Aortic dilatation increases the risk that the aorta will tear (aortic dissection) or rupture, causing serious heart problems or sometimes sudden death. Sometimes, defects in heart valves can also cause problems. In some cases, certain

valves may leak, creating a "heart murmur," which a doctor can hear with a stethoscope. Small leaks may not result in any symptoms, but larger ones may cause shortness of breath, fatigue, and palpitations (a very fast or irregular heart rate).

- **Nervous system.** The brain and spinal cord are surrounded by fluid contained by a membrane called the dura, which is composed of connective tissue. As someone with Marfan syndrome gets older, the dura often weakens and stretches, then begins to weigh on the vertebrae in the lower spine and wear away the bone surrounding the spinal cord. This is called dural ectasia. These changes may cause only mild discomfort; or they may lead to radiated pain in the abdomen; or to pain, numbness, or weakness in the legs.
- **Skin.** Many people with Marfan syndrome develop stretch marks on their skin, even without any weight change. These stretch marks can occur at any age and pose no health risk. However, people with Marfan syndrome are also at increased risk for developing an abdominal or inguinal hernia, in which a bulge develops that contains part of the intestines.
- **Lungs.** Although connective tissue problems make the tiny air sacs within the lungs less elastic, people with Marfan syndrome generally do not experience noticeable problems with their lungs. If, however, these tiny air sacs become stretched or swollen, the risk of lung collapse may increase. Rarely, people with Marfan syndrome may have sleep-related breathing disorders such as snoring, or sleep apnea (which is characterized by brief periods when breathing stops).

What Causes Marfan Syndrome?

Marfan syndrome is caused by a defect, or mutation, in the gene that determines the structure of fibrillin-1, a protein that is an important part of connective tissue. A person with Marfan syndrome is born with the disorder, even though it may not be diagnosed until later in life.

The defective gene that causes Marfan syndrome can be inherited: The child of a person who has Marfan syndrome has a 50 percent chance of inheriting the disease. Sometimes a new gene defect occurs during the formation of sperm or egg cells, making it possible for two parents without the disease to have a child with the disease. But this is rare. Two unaffected parents have only a 1 in 10,000 chance of having a child with Marfan syndrome. Possibly 25 percent of cases are due to a spontaneous mutation at the time of conception.

Although everyone with Marfan syndrome has a defect in the same gene, different mutations are found in different families, and not everyone experiences the same characteristics to the same degree. In other words, the defective gene expresses itself in different ways in different people. This phenomena is known as variable expression. Scientists do not yet understand why variable expression occurs in people with Marfan syndrome.

How Is Marfan Syndrome Diagnosed?

There is no specific laboratory test, such as a blood test or skin biopsy, to diagnose Marfan syndrome. The doctor and/or geneticist (a doctor with special knowledge about inherited diseases) relies on observation and a complete medical history, including:

- Information about any family members who may have the disorder or who had an early, unexplained, heart-related death.
- A thorough physical examination, including an evaluation of the skeletal frame for the ratio of arm/leg size to trunk size.
- An eye examination, including a "slit lamp" evaluation.
- Heart tests such as an echocardiogram (a test that uses ultrasound waves to examine the heart and aorta).

The doctor may diagnose Marfan syndrome if the patient has a family history of the disease, and if there are specific problems in at least two of the body systems known to be affected. For a patient with no family history of the disease, at least three body systems must be affected before a diagnosis is made. Moreover, two of the systems must show clear signs that

are relatively specific for Marfan syndrome.

In some cases, a genetic analysis may be useful in making a diagnosis of Marfan syndrome, but such analyses are often time consuming and may not provide any additional helpful information. Family members of a person diagnosed with Marfan syndrome should not assume they are not affected if there is no knowledge that the disorder existed in previous generations of the family. After a clinical diagnosis of a family member, a genetic study might identify the specific mutation for which a test can be performed to determine if other family members are affected.

Recently, doctors discovered a connective tissue disorder known as Loeys-Dietz syndrome, which has several characteristics that overlap with those of Marfan syndrome. When making a diagnosis, it is important to distinguish between the two disorders: Loeys-Dietz is more likely to cause fatal aortic aneurysms, and treatment for the two is different. A diagnostic test for Loeys-Dietz syndrome is available.

What Types of Doctors Treat Marfan Syndrome?

Because a number of body systems may be affected, a person with Marfan syndrome should be cared for by several different types of doctors. A general practitioner or pediatrician may oversee routine health care and refer the patient to specialists such as a cardiologist (a doctor who specializes in heart disorders), an orthopaedist (a doctor who specializes in bones), or an ophthalmologist (a doctor who specializes in eye disorders), as needed. Some people with Marfan syndrome also go to a geneticist.

What Treatment Options Are Available?

There is no cure for Marfan syndrome. To develop one, scientists may have to identify and change the specific gene responsible for the disorder before birth. However, a range of treatment options can minimize and sometimes prevent complications. The appropriate specialists will develop an individualized treatment program; the approach the doctors use depends on which systems have been affected.

- **Skeletal.** Annual evaluations are important to detect any changes in the

spine or sternum. This is particularly important in times of rapid growth, such as adolescence. A serious malformation not only can be disfiguring, but also can prevent the heart and lungs from functioning properly. In some cases, an orthopaedic brace or surgery may be recommended to limit damage and disfigurement.

- **Eyes.** Early, regular eye examinations are essential for identifying and correcting any vision problems associated with Marfan syndrome. In most cases, eyeglasses or contact lenses can correct the problem, although surgery may be necessary in some cases.
- **Heart and blood vessels.** Regular checkups and echocardiograms help the doctor evaluate the size of the aorta and the way the heart is working. The earlier a potential problem is identified and treated, the lower the risk of life-threatening complications. Those with heart problems are encouraged to wear a medical alert bracelet and to go to the emergency room if they experience chest, back, or abdominal pain. Some heart-valve problems can be managed with drugs such as beta-blockers, which may help decrease stress on the aorta. In other cases, surgery to replace a valve or repair the aorta may be necessary.

Surgery should be performed before the aorta reaches a size that puts it at high risk for tear or rupture. Because blood clots can form around artificial heart valves, people who have a valve replaced must take the blood-thinning drug warfarin for the rest of their lives. They must also take extreme care to prevent endocarditis (inflammation of the lining of the heart cavity and valves). Dentists should be alerted to this risk; they are likely to recommend that the patient be prescribed protective medicines before they perform dental work.

Because warfarin carries a risk of some serious side effects, including excessive bleeding, and because it is dangerous to unborn babies, doctors are increasingly opting for a newer aortic root replacement procedure that enables people to keep their own valves. The procedure involves removing and

replacing the enlarged part of the aorta with a Dacron tube, and resuspending the natural valve into the tube so that the tube supports the valve. The procedure is often performed at an earlier stage than traditional valve replacement. It may also be offered to women with aortic enlargement who are considering becoming pregnant, because it can prevent the rapid aortic growth and possible tearing that sometimes occur during pregnancy.

- **Nervous system.** If dural ectasia (swelling of the covering of the spinal cord) develops, medication may help minimize any associated pain.
- **Lungs.** It is especially important that people with Marfan syndrome not smoke, as they are already at increased risk for lung damage. Any problems with breathing during sleep should be assessed by a doctor.

Pregnancy poses a particular concern due to the stress on the body, particularly the heart. A pregnancy should be undertaken only under conditions specified by obstetricians and other specialists familiar with Marfan syndrome. In some cases, valve surgery prior to pregnancy may be warranted. The pregnancy should be monitored as a high-risk condition. Women with Marfan syndrome may also seek genetic counseling concerning the likelihood that they will pass the disease on to their children.

Although eating a balanced diet is important for maintaining a healthy lifestyle, no vitamin or dietary supplement has been shown to help slow, cure, or prevent Marfan syndrome.

For most people with Marfan syndrome, engaging in moderate aerobic exercise is important for promoting skeletal and cardiovascular health and a sense of well-being. However, because of the risk of aortic dissection, people with the syndrome should not engage in contact sports, competitive athletics, or isometric exercise.

What Are Some of the Emotional and Psychological Effects of Marfan Syndrome?

Being diagnosed and learning to live with a genetic disorder can cause social, emotional, and financial stress. It often requires a great deal of adjustment in outlook and lifestyle. A person who is an adult when Marfan syndrome is diagnosed may feel angry or afraid. There

may also be concerns about passing the disorder to future generations or about its physical, emotional, and financial implications.

The parents and siblings of a child diagnosed with Marfan syndrome may feel sadness, anger, and guilt. It is important for parents to know that nothing that they did caused the fibrillin-1 gene to mutate. Parents may be concerned about the genetic implications for siblings or have questions about the risk to future children.

Some children with Marfan syndrome are advised to restrict their activities. This may require a lifestyle adjustment that is hard for a child to understand or accept.

For both children and adults, appropriate medical care, accurate information, and social support make it easier to live with the disease. Genetic counseling may also be helpful for understanding the disease and its potential impact on future generations.

While Marfan syndrome is a lifelong disorder, the outlook has improved in recent years. As early as the 1970s, the life expectancy of a person with Marfan syndrome was two-thirds that of a person without the disease; however, with improvements in recognition and treatment, people with Marfan syndrome now have a life expectancy similar to that of the average person.

What Research Is Being Conducted on Marfan Syndrome?

Several years ago, National Institutes of Health (NIH)-funded researchers found that losartan, a common blood pressure medication, also affected a molecular pathway involved in aortic weakening and rupture, an often fatal complication of Marfan syndrome.

Numerous studies are underway that should lead to a better understanding of Marfan syndrome and its treatment. They include a plan to identify the factors responsible for the cardiovascular manifestations of Marfan syndrome, a study to better understand the process that leads to skeletal manifestations, and studies to clarify the role of a chemical messenger called transforming growth factor-beta (TGF- β) in the disorder. Some of the most recent studies are identifying new potential therapeutic targets for Marfan syndrome.

Scientists are conducting research on Marfan syndrome from a variety of perspectives. One approach is to better understand what happens once the genetic defect or mutation occurs. How does it change the way connective tissue develops and functions in the body? Why are people with Marfan syndrome affected differently? Scientists are searching for the answers to these questions both by studying the genes themselves and by studying large family groups affected by the disease. Mouse models that carry mutations in the fibrillin-1 gene may help scientists better understand the disorder. Animal studies that can provide preliminary information for gene therapy are also underway.

Other scientists are focusing on ways to treat some of the complications that arise in people with Marfan syndrome. Clinical studies are being conducted to evaluate the usefulness of certain medications in preventing or reducing problems with the aorta.

For example, research has shown that the blood pressure medication losartan prevents aortic aneurysms in a mouse model of Marfan syndrome. Studies receiving funding are now underway to determine whether the drug has the same beneficial effect in people.

More information on research is available from the following websites:

- [NIH Clinical Research Trials and You](#) helps people learn more about clinical trials, why they matter, and how to participate. Visitors to the website will find information about the basics of participating in a clinical trial, first-hand stories from actual clinical trial volunteers, explanations from researchers, and links to how to search for a trial or enroll in a research-matching program.
- [ClinicalTrials.gov](#) offers up-to-date information for locating federally and privately supported clinical trials for a wide range of diseases and conditions.
- [NIH RePORTER](#) is an electronic tool that allows users to search a repository of both intramural and extramural NIH-funded research projects from the past 25 years and access publications (since 1985) and patents resulting from NIH funding.
- [PubMed](#) is a free service of the U.S.

National Library of Medicine that lets you search millions of journal citations and abstracts in the fields of medicine, nursing, dentistry, veterinary medicine, the health care system, and preclinical sciences.

Where Can People Find More Information About Marfan Syndrome?

National Institute of Arthritis and Musculoskeletal and Skin Diseases (NIAMS) Information Clearinghouse National Institutes of Health

1 AMS Circle
Bethesda, MD 20892-0001
Phone: 301-495-4484
Toll Free: 877-22-NIAMS (877-226-4267)
TTY: 301-565-2966
Fax: 301-718-6366
Email: NIAMSinfo@mail.nih.gov
Website: <http://www.niams.nih.gov>

Other Resources

National Human Genome Research Institute (NHGRI) National Institutes of Health

Website: <http://www.genome.gov>

NHGRI Genetic and Rare Diseases Information Center

Website: <http://www.genome.gov>

National Marfan Foundation (NMF)

Website: <http://www.marfan.org>

National Organization for Rare Disorders (NORD)

Website: <http://www.rarediseases.org>

March of Dimes Birth Defects Foundation

Website: <http://www.marchofdimes.com>

American Heart Association

Website: <http://www.americanheart.org>

For additional contact information, visit the NIAMS website or call the NIAMS Information Clearinghouse.

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For Your Information

This publication contains information about medications used to treat the health condition discussed here. When this booklet was printed, we included the most up-to-date (accurate) information available. Occasionally, new information on medication is released.

For updates and for any questions about any medications you are taking, please contact

U.S. Food and Drug Administration

Toll Free: 888-INFO-FDA
(888-463-6332)
Website: <http://www.fda.gov>

For updates and questions about statistics, please contact

Centers for Disease Control and Prevention's National Center for Health Statistics

Toll Free: 800-232-4636
Website: <http://www.cdc.gov/nchs>

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